

What is claimed is:

1. An isolated nucleic acid molecule that comprises at least 20 consecutive nucleotides but not more than 1500 consecutive nucleotides of the sequence of SEQ ID NO: 1.
2. An isolated nucleic acid molecule comprising a promoter which comprises at least 20 consecutive nucleotides but not more than 1500 consecutive nucleotides of the sequence of SEQ ID NO: 1, said promoter being operably linked to a heterologous nucleic acid sequence.
3. The isolated nucleic acid molecule according to claim 2, where said heterologous nucleic acid sequence is capable of being expressed in ocular tissue.
4. The isolated nucleic acid molecule according to claim 2, where said heterologous nucleic acid sequence is capable of being expressed in optic nerve cells.
5. The isolated nucleic acid molecule according to claim 2, where said heterologous nucleic acid sequence is capable of being expressed in retinal cells.
6. The isolated nucleic acid molecule according to claim 2, where said heterologous nucleic acid sequence is capable of being expressed in trabecular meshwork cells.

7. The isolated nucleic acid molecule according to claim 2, where said heterologous nucleic acid sequence is selected from the group consisting of a coding sequence, a toxin, and a reporter gene.

8. The isolated nucleic acid molecule according to claim 7, wherein the reporter gene is selected from the group consisting of green fluorescent protein and luciferase.

9. The isolated nucleic acid molecule according to claim 2, where said heterologous nucleic acid sequence is capable of being transcribed as an antisense RNA.

10. The isolated nucleic acid molecule according to claim 9, wherein said antisense RNA is capable of binding to a nucleic acid molecule having the nucleotide sequence of SEQ ID NO: 1 or complements thereof under physiological conditions.

11. The isolated nucleic acid molecule according to claim 10, wherein said antisense RNA is capable of binding to a nucleic acid molecule having a nucleotide sequence selected from the group consisting of SEQ ID NOs: 3 through 463 and complements thereof under physiological conditions.

12. A nucleic acid molecule capable of detecting a single nucleotide polymorphism selected from table 1.

13. The nucleic acid molecule according to claim 12, wherein the nucleic acid molecule is capable of detecting a single nucleotide polymorphism selected from table 4.

14. The nucleic acid molecule according to claim 12, wherein said nucleic acid molecule is capable of detecting a guanine.

15. The nucleic acid molecule according to claim 12, wherein said nucleic acid molecule is capable of detecting a cytosine.

16. The nucleic acid molecule according to claim 12, wherein said nucleic acid molecule is capable of detecting a thymine.

17. The nucleic acid molecule according to claim 12, wherein said nucleic acid molecule is capable of detecting an adenine.

18. The nucleic acid molecule according to claim 12, wherein said nucleic acid molecule does not specifically hybridize to a nucleic acid molecule consisting of SEQ ID NO: 1.

19. A nucleic acid molecule capable of detecting a single nucleotide polymorphism in an optineurin promoter by specifically detecting said single nucleotide polymorphism in said optineurin promoter, wherein said nucleic acid molecule does not specifically hybridize to a nucleic acid molecule consisting of SEQ ID NO: 1.

20. A host cell comprising a nucleic acid molecule comprising a promoter which comprises at least 20 consecutive nucleotides but not more than 1500 consecutive nucleotides of the sequence of SEQ ID NO: 1, said promoter being operably linked to a heterologous nucleic acid sequence.

21. The host cell of claim 20, wherein said host cell is selected from the group consisting of a non-human mammalian cell, a bacterial cell, and an isolated human cell.

22. A method for diagnosing glaucoma in a sample obtained from a cell or a bodily fluid by detecting a polymorphism in a promoter region of the optineurin gene, comprising the steps of:

(A) incubating under conditions permitting nucleic acid hybridization, a marker nucleic acid molecule, said marker nucleic acid molecule having a nucleic acid sequence that specifically hybridizes to a sequence selected from the group consisting of SEQ ID NO: 1 and a complement thereof, and a complementary nucleic acid molecule obtained from a sample, wherein nucleic acid hybridization between said marker nucleic acid molecule and said complementary nucleic acid molecule permits the detection of said polymorphism;

(B) permitting hybridization between said marker nucleic acid molecule and said complementary nucleic acid molecule; and

(C) detecting the presence of said polymorphism, wherein the detection of said polymorphism is diagnostic of glaucoma.

23. The method for diagnosing glaucoma of claim 22, wherein said polymorphism is a single nucleotide polymorphism.

24. The method for diagnosing glaucoma of claim 22, wherein said marker nucleic acid molecule has a nucleotide sequence selected from the group consisting of SEQ ID NOs: 3 through 463.

25. The method for diagnosing glaucoma of claim 22, further comprising a second marker nucleic acid molecule.

26. The method for diagnosing glaucoma of claim 22, wherein the cell or bodily fluid comprises ocular tissue.

27. The method for diagnosing glaucoma of claim 22, wherein the cell or bodily fluid comprises optic nerve cells.

28. The method for diagnosing glaucoma of claim 22, wherein the cell or bodily fluid comprises retinal cells.

29. The method for diagnosing glaucoma of claim 22, wherein the cell or bodily fluid comprises a bodily fluid selected from the group consisting of glaucomatous cell extract, fluid from the anterior chamber of the eye, blood, lymph, and serum.

30. The method for diagnosing glaucoma of claim 22, further comprising amplifying the complementary nucleic acid molecule obtained from a sample using a nucleic acid amplification method.

31. The method for diagnosing glaucoma of claim 22, wherein the nucleic acid amplification method is selected from the group consisting of polymerase chain amplification, ligase chain reaction, oligonucleotide ligation assay, thermal amplification, and transcription base amplification.

32. A method for prognosing glaucoma in a sample obtained from a cell or a bodily fluid by detecting a polymorphism in a promoter region of the optineurin gene, comprising the steps of:

(A) incubating under conditions permitting nucleic acid hybridization, a marker nucleic acid molecule, said marker nucleic acid molecule having a nucleic acid sequence that specifically hybridizes to a sequence selected from the group consisting of SEQ ID NO: 1 and complement thereof, and a complementary nucleic acid molecule obtained from a sample, wherein nucleic acid hybridization between said marker nucleic acid molecule and said complementary nucleic acid molecule permits the detection of said polymorphism;

(B) permitting hybridization between said marker nucleic acid molecule and said complementary nucleic acid molecule; and

(C) detecting the presence of said polymorphism, wherein the detection of said polymorphism is prognostic of glaucoma.

33. The method for prognosing glaucoma of claim 32, wherein said polymorphism is a single nucleotide polymorphism.

34. The method for prognosing glaucoma of claim 32, wherein said marker nucleic acid molecule has a nucleotide sequence selected from the group consisting of SEQ ID NOs: 3 through 463.

35. The method for prognosing glaucoma of claim 32, further comprising a second marker nucleic acid molecule.

36. A method for diagnosing or prognosing glaucoma in a sample obtained from a cell or a bodily fluid by detecting a polymorphism in a promoter region of the optineurin gene, comprising the steps of:

(A) incubating under conditions permitting nucleic acid hybridization, a marker nucleic acid molecule, said marker nucleic acid molecule having a nucleic acid sequence that specifically hybridizes to a optineurin promoter sequence or its complement, and a complementary nucleic acid molecule obtained from a sample, wherein nucleic acid hybridization between said marker nucleic acid molecule and said complementary nucleic acid molecule permits the detection of said polymorphism;

(B) permitting hybridization between said marker nucleic acid molecule and said complementary nucleic acid molecule; and

(C) detecting the presence of said polymorphism, wherein the detection of said polymorphism is diagnostic or prognostic of glaucoma.

37. The method for diagnosing or prognosing glaucoma of claim 36, wherein said optineurin promoter sequence comprises SEQ ID NO: 1 or a fragment thereof.

38. The method for diagnosing or prognosing glaucoma of claim 36, wherein said marker nucleic acid is capable of specifically detecting a single nucleotide polymorphism.

39. The method for diagnosing or prognosing glaucoma of claim 36, wherein said marker nucleic acid molecule has a nucleotide sequence selected from the group consisting of SEQ ID NOs: 3 through 463.

40. The method for diagnosing or prognosing glaucoma of claim 36, further comprising a second marker nucleic acid molecule.

41. A method for detecting the presence or absence of a SNP sequence variation in a sample containing DNA, comprising contacting a labeled nucleic acid capable of detecting a single nucleotide polymorphism selected from table 1 with the DNA of the sample under hybridization conditions and determining the presence of hybrid nucleic acid molecules comprising the labeled nucleic acid.

42. The method of claim 41, wherein the sample containing DNA is derived from a human with elevated intraocular pressure.

43. The method of claim 41, wherein the sample containing DNA is derived from a human without elevated intraocular pressure.

44. A method for detecting the presence or absence of an optineurin promoter sequence variation in a sample containing DNA, comprising providing amplification reaction primers that direct amplification of a selected nucleic acid region containing said sequence variation within said optineurin promoter, amplifying the nucleic acid defined by the amplification reaction primers, and determining the presence or absence of said sequence variation.

45. The method of claim 44, wherein the determining the presence or absence of said sequence variation comprises sequencing the amplified nucleic acid.



46. The method of claim 44, wherein the determining the presence or absence of said sequence variation comprises a hybridization assay.

47. A method for determining the presence of increased susceptibility to a glaucoma, or to a progressive ocular hypertensive disorder resulting in loss of visual field in a patient, or the severity or progression of glaucoma in a patient, comprising providing amplification reaction primers that direct amplification of a selected nucleic acid region containing said sequence variation within said optineurin promoter, amplifying the nucleic acid defined by the amplification reaction primers, and determining the presence or absence of said sequence variation.

48. A method for detecting a polymorphism comprising: obtaining a sample containing human genomic DNA, providing a nucleic acid molecule capable of detecting a single nucleotide polymorphism located with an optineurin promoter, and detecting the presence or absence of said polymorphism.

49. The method detecting a polymorphism according to claim 48, wherein said polymorphism is selected from table 1.

50. A kit for determining the presence of increased susceptibility to a glaucoma, or to a progressive ocular hypertensive disorder resulting in loss of visual field, or the severity or progression of glaucoma in a patient, comprising a labeled nucleic acid capable of detecting a single nucleotide polymorphism selected from table 1 and a means

for detecting hybridization with the labeled nucleic acid, and instructions for using said kit.

51. A kit for determining the presence of increased susceptibility to a glaucoma, or to a progressive ocular hypertensive disorder resulting in loss of visual field in a patient, or the severity or progression of glaucoma in a patient, comprising amplification reaction primers that direct amplification of a selected nucleic acid region containing a characteristic nucleotide of an optineurin promoter SNP sequence variant and an enzyme for amplifying the region containing said characteristic nucleotide.